Congenital ichtyosis – a clinical research

Overview. Congenital ichtyosis represents a heterogenous group of desquamative skin disorders among which the most frequent are ichtysosis vulgaris and X-linkate ichtyosis.

Material. 111 medical files of patients with congenital ichtysosis, who have been treated at the Hospital of Dermatology and Communicable Diaseases during 1986-2015, were included in the reasearch.

Results. Patients' age has oscillated from 1 till 18 years old (m/f-42/69; rural/urban – 87/24). Range of clinical forms included, as follows: ichtyosis vulgaris – 76 (68.5%) cases; non-bulous ichtyoziform erythroderma – 23 (20.7%) cases; bullous ichtyoziform erythroderma of Brocq – 3 (2.7%) cases; X-linkate ichtyosis – 7 (6.3%) cases; Netherton syndrome – 2 (1.8%) cases. Thus, autosomal-dominant transmission of the disease was established in 79 (71.2%) cases, as well as autosomal-recessive was present in 25 (22.5%) cases and X-linkate in 7 (6.3%) cases. All patients prior have developed recurrent episodes of amigdalitis, bronchitis, pneumonia, acute viral infections, pyodermas and anemia. Both, somatic and mental retardation, were seen in 32 (28.8%) cases. Benign forms of ichtyosis vulgaris were described as follows: simple form in 70 cases; xeroderma and nigricans form 3 cases of each. Onset of the disease between 2 and 12 months old was observed in 53 (69.7%) cases, from 1 till 4 years old in 17 (22.3%) cases and after 4 years in 6 (8%) cases. In majority of cases, a desquamation with fine white scales on extensor surface of limbs, less on trunk, face and scalp of the patients were seen; nigricans form was an exception. Also, a massive hyperkeratotic depositions on patients' elbows and knees were present, as well as follicular hyperkeratosis with accentuated skin markings on palms and soles were observed in 51 (67.1%) cases. Cases of X-linkate ichtyosis have been chracterized by an early onset within first 6 months of patients' life, a benign evolution of the disease, large polygonal scales, absence of follicular hyperkeratosis and involvement of palms and soles. Onset of ichtyosiform erythroderma (recessive non-bullous and dominant bullous forms) was described at delivery with "collodion bebe" manifesations, as well as benign evolution, in majority of cases, on the 10-14th days of life. A severe progression of the disease with generalized skin involvement was seen in 18 (69.2%) cases, associated with onichodystrophy and trichodistrophy in a half of cases. It is important to notice an extreme severe evolution of bullous ichtyosiform erythroderma in 3 children with early onset, bulla and wide exfoliation of the epidermis. Late clinical manifestations were presented by multilayered, verrucose, gray-brown scales, which covered a generalized erythema; palmo-plantar keratoderma, onicodystrophy, alopecia and pyodermas were seen as well. Development of Netherton syndrome was established in the first 3-4 months of patients' life. Clinical aspects included as follows: liniar ichtyosis Comel, atopic eczema and trichodystrophy (hypotrichosis and trichorrhexis invaginata). Topical treatment has involved remedies with urea, salicylic acid, emolients and vitamin A with a good effect in patients with ichtyosis vulgaris (xeroderma, simple form) and a temporary benefic result in other forms.

Conclusions. Ichtyosis occurs mostly in females; prevalence of dominant forms with a benign evolution and positive response to topical treatment were observed; only bullous ichtyosiform erythroderma of Brocq made an exception. Ressecive forms of ichtyosis were marked by severe evolution and treatment resistance. Consultation of a genetic specialist repersents a unique solution for antenatal prohylaxis of the disease.

Key words: congenital ichtyosis, children, particularities.

Epidermolizele buloase – studiu clinic

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Generalități. Au fost analizate fișele de observație clinică (41 de cazuri) ale pacienților cu epidermolize buloase (EB), tratați în perioada 2000-2014, în cadrul Spitalului Dermatologie și Maladii Comunicabile.

Rezultate obținute. Vârsta pacienților a oscilat între 2 luni și 52 de ani (m/f –28/13; rural/urban – 25/16). Spectrul nosologic depistat, s-a prezentat în felul următor: epidermoliza buloasă simplă (EBS) Koebner – 11 cazuri; EBS Weber–Cockayne – 9 cazuri; EBS asociată cu tulburări de pigmentare – 1 caz; epidermoliza buloasă joncțională (EBJ) Herlitz – 1 caz; epidermoliza buloasă distrofică dominantă (EBDD) Cockayne-Touraine – 12 cazuri; EBDD Passini – 3 cazuri; epidermoliza buloasă distrofică recesivă (EBDR) Hallopeau–Siemens – 4 cazuri. Așadar, EBS s-a constatat în 51,2% cazuri, EBJ – în 2,4% și EBD – în 46,34% cazuri. Manifestările buloase de epidermoliză au fost asociate cu o afectare unghială în 48,8% cazuri. Hiperhidroza, keratodermia palmo-plantară, alopecia și leziunile ichtioziforme s-au evidențiat în 12,2% cazuri. La circa o jumătate dintre pacienți au fost constatate o serie de maladii concomitente (anemii, pneumonii, hepatite, pancreatite, pielonefrite). Anomaliile dentare și afectarea mucoaselor s-au raportat la o treime dintre pacienți, fiecare al zecelea pacient având și retard mental. La circa 20% cazuri, au fost observate complicații cu agenți microbieni (piodermii), iar la bolnavii cu EBDR Hallopeau–Siemens – cheloizi, acrosclerodactilie, contracturi, mutilații, stenoze esofagiene. Un caz de deces s-a constatat la pacientul cu EBJ Herlitz, la 2 luni de la naștere. O evoluție favorabilă s-a observat la pacienții cu forme epidermolitice și distrofice dominante (63,4%). Pe durata tratamentului de rutină, s-a constatat apariția bulelor noi la toți bolnavii. Epitelizarea eroziunilor s-a observat pe parcursul a 5-14 zile. După includerea în tratamentul topic a pansamentelor „Mepilex Lite", „Mepitel", durata epitelizării erupției s-a redus de 2 ori (3-7 zile).

Concluzii. Profilul epidermolizei buloase constată predominarea sexului masculin, precum și a formelor clinice de EB dominante; asocierea cu afectările unghiale și a mucoaselor, cu distrofii dentare, anemii, complicații microbiene, în cazuri grave – acrosclerodactilie, mutilații, stenoză esofagiană. Includerea în tratamentul topic a pansamentelor „Mepilex Lite", „Mepitel" reduce durata epitelizării eroziunilor.

Cuvinte-cheie: epidermolize buloase, particularități clinice, dificultăți de tratament.

Epidermolysis bullosa – a clinical study

Overview. The study included case histories of 41 patients with epidermolysis bullosa, who were treated in Hospital of Dermatology and Communicable Diseases during 2000-2014.

Results. Patients' age has oscillated from 2 till 52 years old (M/F-28/13; Rural/Urban – 25/16). Nosological spectrum ranged as follows: simple epidermolysis bullosa (EBS) Koebner–11 cases; EBS Weber-Cockayne – 9 cases; EBS associated with pigmented lesions – 1; jonctional epidermolysis bullosa – (EBJ) Herlitz – 1; dystrophic dominant form of EB (EBDD) Cockayne-Touraine – 12; EBDD Passini – 3; recessive form of EBD (EBDR) Hallopeau – Siemens – 4. Thus, EBS has been established in 51.2% of cases, EBJ – 2.4% of cases and EBD – in 46.34% of cases. Nail involvement was observed in 48.8 cases of EB. Hyperhidrosis, palmoplantar keratoderma, alopecia and ichthyosiform lesions were marked in 12.2% of cases. About half of patients has manifested a plenty number of associated diseases (anemia, pneumonia, hepatitis, pancreatitis, pyelonephritis). Dental anomalies and mucous membranes involvement were reported in 1/3 of patients, one in every 10 patients presented a mental retardation. In 20% of cases pyococcal complications (pyodermas) was described, in addition patients with EBDR Hallopeau-Siemens have developed keloid scars, acrosclerodactylitis, muscular contractions, mutilations, esophageal stenosis. A 2 month old infant with EBJ-H has died. More benign evolution was marked in patients with dominant forms of EBD (63.4%). All patients have presented a new bulla during obvious therapy. Epithelization of erosions occurred within 5-14 days. Topical treatment with "Mepilex Lite", "Mepitel" reduced twice the time of lesion epithelization (3-7days).

Conclusions. Epidermolysis bullosa profile certifies prevalence of this disease among males, as well as, increased number of dominant forms, association with nail and mucous membrane involvement, dental anomalies, anemia, microbial infections, in severe cases – acroscelro-dactylitis, mutilations, esophageal stenosis. "Mepilex Lite" and "Mepitel" dressings have reduced the time of lesion epithelization.

Key words: epidermolysis bullosa, clinical features, treatment difficulties.

Pemfigus benign familial Hailey-Hailey – prezentare de caz

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Generalități. Boala Hailey-Hailey (pemfigus benign familial) este o genodermatoză (mutații în gena ATP2C1) rară, cu transmitere autosomal-dominantă, caracterizată printr-o fragilitate crescută a pielii și structură anormală a desmozomilor. La instalarea diagnosticului, pe lângă tabloul clinic specific și antecedentele familiale, o importanță semnificativă o are examenul histopatologic: bula situată suprabazal, papilomatoza – papile dermice cu aspect „în degete de mănușă", care proemină în interiorul bulei, pe alocuri cu asociere de hiperkeratoză.

Prezentare de caz. Prezentăm cazul unei paciente de 43 de ani, din mediul rural, observată în secția femei a Spitalului Dermatologie și Maladii Comunicabile, cu diagnosticul *Pemfigus* benign familial Hailey-Hailey, durata maladiei fiind de aproximativ 7 ani. Pacienta s-a tratat ambulator, de mai multe ori, pentru intertrigo și lichen plan, cu efect curativ neînsemnat. Cazuri asemănătoare s-au constatat la bunică, tată, soră și frate. Pentru prima dată, erupția s-a observat pe gât. Datele obiective relevă prezența plăcilor eritemato-veziculo-buloase, zemuinde, circinate, cu tendință spre confluare, macule hipercromice, localizate în pliurile axilare, submamare și pe gât, asociate de prurit. Semnul Nikolsky a fost negativ. Diagnosticul s-a stabilit în baza următoarelor date: tablou clinic, antecedente familiale, examen histopatologic. S-a observat o evoluție favorabilă în tratamentul indicat, care a inclus: corticoizi pe cale generală (doze moderate de prednison cu reducere treptată), dermatocorticoizi, loțiuni antiseptice.

Concluzii. Cazul este adus în discuție pentru raritatea acestuia și particularitățile clinico-evolutive specifice. Maladia a evoluat timp de mai mulți ani cu aspect de intertrigo și lichen plan, ceea ce a creat dificultăți de diagnostic pozitiv și diferențial.

Cuvinte-cheie: *pemfigus* Hailey-Hailey, genodermatoză rară, prezentare de caz.

Familial benign pemphigus Hailey-Hailey: a case presentation

Overview. Hailey-Hailey disease (familial benign pemphigus) is rare autosomal-dominant genodermatoses (induced by a mutation in ATP2C1 gene), which is chracterized by an increased skin fragility and abnormal structure of the desmosomes.

A case report. A 43 year old female patient, originated from a rural region,was treated in Hospital of Dermatology and Communicable Diseases for familial benign pemphigus,the disease lasted for approximately 7 years. Initially,the patient was treated for intertrigo and lichen planus, but without any significant effect. Similar clinical manifestations have been seen in the patient's relatives: grandmother, father, sister and brother. For the first time, skin lesions occurred on the patients' neck. Clinical inspection revelead presence of erythematous macules covered with vesicle, bulla and erosions with oozing, which had a tendency to confluent together, itching hyperpigemented macules were seen in axila, submamary folds, as well as on neck. Nicolsky sign was negative. Diagnosis was made on the basis of clinical signs, heredetary anamnesis and histopathological report. Treament, which included systemic steroids, moderate dosage of prednisolone, topical steroids and antiseptic lotions, has shown a positive result.

Conclusion. This case was presented for discussion due to its rarity.

Key words: Hailey-Hailey disease, rare genodermatosis, case report.